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Mixed gonadal dysgenesis with an ovotestis on imaging mimicking ovotesticular disorder of sexual differentiation

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ABSTRACT

Mixed gonadal dysgenesis (MGD) is a rare disorder of sexual development. Also known as 45XO/46XY mosaicism, MGD is characterized by highly variable sexual phenotypes and an increased risk of gonadal malignancy. Patients with MGD often have a unilateral descended gonad and contralaterally either a streak gonad or no gonad. We present the case of a patient with a dysgenetic, nonpalpable gonad with imaging features of an ovotestis. These imaging features are generally more indicative of ovotesticular disorder of sexual development (previously true hermaphrodite), which is a condition with low risk of gonadal malignancy. Further evaluation with histology and genetic analysis confirmed the diagnosis of MGD. It is important to diagnose MGD to allow for early operative intervention and screening for malignancy.

KEYWORDS 45XO/46XY mosaicism; disorders of sexual development; mixed gonadal dysgenesis; ovotestis

The clinical presentation of mixed gonadal dysgenesis (MGD) varies widely and may be phenotypic female, male, or ambiguous. Most individuals with MGD have a unilateral descended gonad and a contralateral streak gonad or no gonad. MGD is associated with infertility, insufficient puberty, and stunted growth in adolescence. There is about a 33% risk of developing malignancy in the dysgenetic gonad in the first two decades if XY chromosomes are present.[1]

CASE REPORT

A 3-month-old infant was referred to urology for hypospadias. Physical examination revealed a normal size phallus, a midscrotal urethral opening, a morphologic left labia, a nonpalpable left gonad, and a palpable gonad in a morphologic right hemiscrotum *(Figure 1)*. Chromosome analysis revealed a mosaic karyotype 45, X/46, XY. Initial ultrasound demonstrated a normal right testicle and fluid collection behind the bladder; magnetic resonance imaging (MRI) revealed a left hemivagina filled with fluid, accounting for the sonographic finding; and fluoroscopic genitography and voiding cystourethrogram confirmed communication with the urethra *(Figure 2)*. A uterine horn extended to the left inguinal canal, where gonadal tissue with imaging characteristics of both testicle and ovary resided *(Figure 3)*.

Left gonad histology confirmed that both testicular and ovarian tissue were present. Ovarian-type stroma contained luteinized stromal cells and rare degenerating follicle-type structures admixed with seminiferous tubules containing germ and Sertoli cells. Definite primordial follicles were not identified, leading to a diagnosis of MGD rather than ovotesticular disorder of sexual development (DSD). Management entailed removing the streak gonad and female anatomical structures, as the parents preferred to raise the child as a male and the external genitalia were relatively masculinized.

DISCUSSION

DSD describes a broad class of conditions defined by atypical chromosomal and anatomical sexual features, with an incidence of 1 in 4500 to 1 in 5000 live births.[1] MGD falls under the umbrella of DSD and is rare, with an incidence of 1.5 in 10,000 births.[2]

Imaging evaluation of patients with suspected DSD includes ultrasound, voiding cystourethrogram, genitography,

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Figure 1. Clinical photograph of the 3-month-old infant demonstrating a right hemiscrotum with palpable gonad, morphologic left labia, normal size phallus, and penoscrotal hypospadias (arrow).

and MRI. These allow for the identification of gonads, internal sex organs, and their anatomical communications. Ultrasound is the first line of imaging, as it is readily available, often adequately defines the anatomy, and does not involve radiation exposure.[3] A voiding cystourethrogram and genitography use fluoroscopic imaging and catheterization to evaluate communications between the internal structures and external genitalia. MRI provides a more detailed view of anatomy if not defined by ultrasound and assists in staging of malignancy when present. Laparoscopy obtains tissue to assess the pathology and histology of the internal sex organs.[4]

This case is unique because the contralateral gonad had the imaging appearance of an ovotestis, and histology was necessary to differentiate MGD from ovotesticular DSD. While both may contain a mixture of male and female sexual features, the gonad does not contain primordial follicles in MGD but they are numerous in ovotesticular DSD.[5] This distinction is important since MGD has a much higher gonadal malignancy risk.[6] Also, it is important to understand that a typical imaging appearance of ovary, testis, or ovotestis may still represent a dysgenetic streak gonad, so biopsy is necessary.[7]

Management includes removal of the streak gonad and biopsy and/or close monitoring of the remaining testicle for

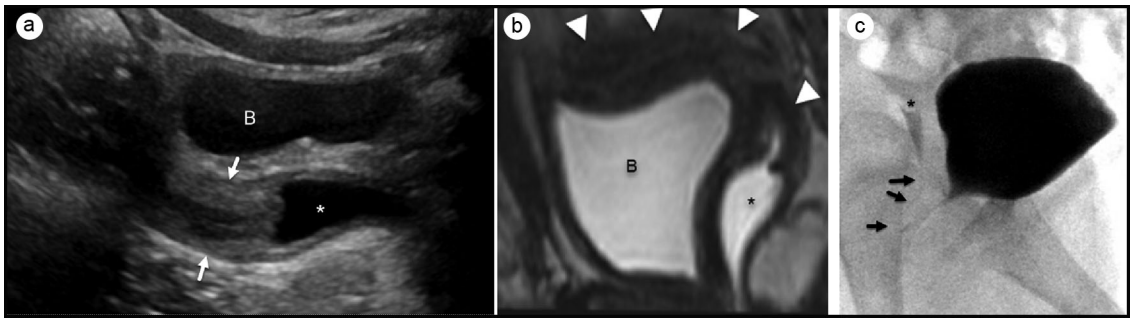

Figure 2. Evaluation of the intrapelvic contents at 3 months of age. **(a)** Initial ultrasound demonstrating a triangular fluid collection (*) posterior to the bladder (B) and adjacent soft tissue echogenicity (arrows) of indeterminate origin. **(b)** Oblique T2 MRI reformatted image demonstrating a rudimentary left uterine horn and cervix (arrows) accounting for the echogenicity on ultrasound, with fluid in a hemivagina (*) posterior to the bladder (B). **(c)** Voiding cystourethrogram near the end of voiding demonstrating communication (arrows) between the posterior urethra and hemivagina with a cervical impression (*).

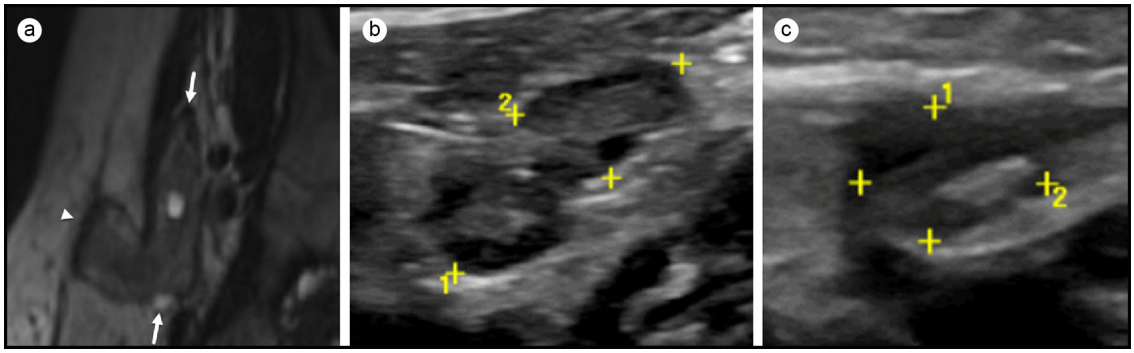

Figure 3. Evaluation of the nonpalpable gonad. **(a)** Double oblique T2 MRI reformatted image showing the ovarian tissue (long arrows) just within the internal ring of the inguinal canal and the testicular tissue (arrowhead) extending into the canal. **(b)** Follow-up targeted ultrasound of the inguinal region confirming the typical sonographic pattern of ovarian stroma and peripheral cysts. **(c)** Follow-up targeted ultrasound of the inguinal region confirming the typical sonographic pattern of hypoechoic testicular parenchyma and hyperechoic mediastinum testis.

malignancy risk. Sex assignment in MGD is complex and depends on multidisciplinary evaluation assessing the degree of virilization, wishes of the family, and psychological and environmental factors.

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Avocations

ONEROUS

I often wonder whatever seized me
When I opted for cardiac bypass.
I knew there is that within me which is
Waning and I bid for Cardiac Cath.
There arose the dilemma of lifetime
Although foggy from sedation, I could
Read furrows of concern on his forehead.
"There is seventy percent narrowing
Of the right coronary." He declared,
In apologetic tone. "Let's fix it,"
I blurted, fighting fog. Then followed
Frenzy of options hurled by doctor friends.
I searched the proven course and I culled the
Anecdotes–stents, drugs, robotic surgery.
In the end, I chose the wonted Bypass.
The lumbering burden was all mine,
A curse of the medical knowledge.

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